

PARENT FACT SHEET

DISORDER

Long Chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)

CAUSE

People with LCHADD have problems breaking down a fatty acid into energy for the body. LCHADD occurs when an enzyme called long chain 3-hydroxyacyl-CoA dehydrogenase is either missing or not working properly. When this enzyme is missing or not working properly the body cannot break down fat for energy so it relies on glucose, which the body only has limited amounts. Once the glucose is used up, the body tries to use fats without success. This leads to low blood sugar and the buildup of harmful substances in the blood.

IF NOT TREATED

The symptoms can vary from person to person. Hypoglycemia (low blood sugar) is usually the first symptom of a metabolic crisis. If the metabolic crisis is not treated, then the baby can have breathing problems, swelling of the brain, seizures and coma, sometimes leading to death.

If not treated the baby may have poor weight gain, delays in learning, delays in walking and other motor skills, enlarged liver, enlarged heart, vision loss, anemia, nerve problems and bouts of muscle weakness and pain.

TREATMENT OPTIONS

- Your doctor will work with a metabolic specialist and dietitian to care for your child.
- Avoid hypoglycemia by having your baby eat at least every 4-6 hours; some babies may need to eat more often. It is important to feed them even during the night. They need to be woken up to eat if they do not wake up on their own.
- A diet high in carbohydrates and low in fat is recommended. Any diet changes should be made under the guidance of a dietitian.
- MCT oil and L-Carnitine are often used as part of the food plan for people with LCHADD. Your metabolic doctor will can guide you on how to use and will give you a prescription for these.
- Call your doctor at the start of any illness.
- Avoid prolonged exercise or exertion.

IF TREATED

Children who are treated early usually live healthy lives with typical growth and development. Some children continue to have episodes of hypoglycemia even with treatment. Some people still have vision, muscle, liver or heart problems even with treatment.